OPHTHALMOLOGY CYSTINOSIS FORUM AFTERNOON SESSION

This symposium took place on 5th December 2016 in Vienna, Austria

Chairpersons

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Speakers

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MEETING SUMMARY

This 1-day meeting was held at the Austria Trend Savoyen Hotel, Vienna, Austria. The afternoon, chaired by Dr Hong Liang and Dr Rachel Bishop, consisted of a series of case presentations focussed on the treatment of ocular manifestations resultant of cystinosis, including the use of the novel gel-like formulation of cysteamine. Dr Inês Leal presented a series of four cases from the Hospital de Santa Maria, Lisbon, Portugal, illustrating the importance of early administration of treatment. Dr Giancarlo Iarossi presented a case highlighting the various examination techniques available to evaluate morphological and functional change during follow-up. Mr Susmito Biswas presented an interesting case of ocular cystinosis with concurrent congenital glaucoma, which brings a number of challenges. Dr Carla Ferreira presented two cases from the Hospital São João, Oporto, Portugal, that had two very different outcomes. Finally, Dr Eleonora Maria Soler Pava presented two cases from her clinic in Bogotá, Colombia, highlighting the good prognosis for patients when continual treatment is available.

Ocular Cystinosis: Clinical Experience in Adult Patients

Doctor Inês Leal

Case 1 reported a 28-year-old female patient diagnosed at the age of 25 years. During investigations for severe hypertension and renal failure during pregnancy the patient was sent to the ophthalmology department for exclusion of hypertensive retinopathy. Corneal crystals were found, suggesting juvenile nephropathic cystinosis, further confirmed by elevated levels of leukocyte cystine. The patient was prescribed systemic and topical cysteamine with visible reduction of corneal crystal deposition by slit lamp biomicroscopy observation 6 months after starting treatment (Figure 1). She is currently on haemodialysis awaiting a renal graft.

Case 2 detailed a 20-year-old male patient diagnosed with infantile nephropathic cystinosis at the age of 1 year. He has been on systemic and topical cysteamine since diagnosis, however there are continued corneal crystals despite years of therapy.

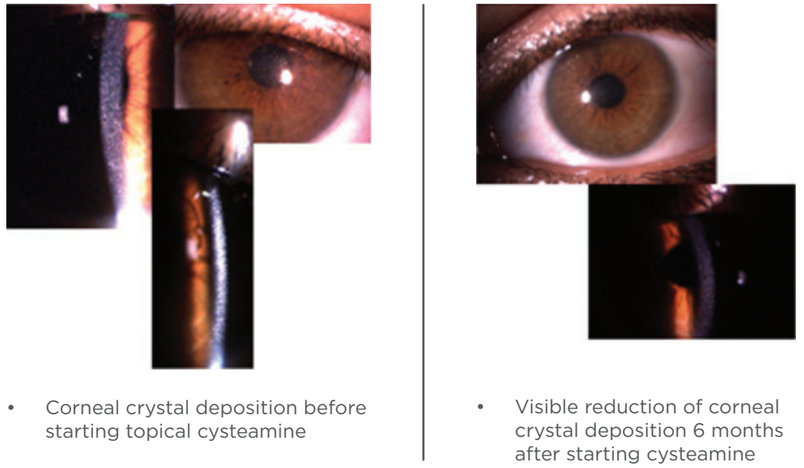
Case 3 described a 42-year-old male patient diagnosed with juvenile nephropathic cystinosis at the age of 41 years. After diagnosis, the patient began systemic and topical cysteamine. Although there has been no obvious decrease in corneal crystal deposition by slit lamp biomicroscopy observation, the patient reports a decrease in photophobia and foreign body sensation, and improved quality of life.

Patients 1, 2, and 3 have had continuous multidisciplinary follow-up since diagnosis. Despite having multiple keratic crystals, visual acuities remain normal and there are no other changes in the complete ophthalmological exam, with no crystals in the iris, angle, or retina.

Case 4 is a 42-year-old female patient diagnosed with infantile nephropathic cystinosis at the age of 2 years. This patient had no regular treatment or follow-up and only started topical cysteamine at the age of 40 years. The patient has bilateral end-stage corneal disease and is legally blind in both eyes.

Three patients have been switched from pharmacy preparation topical cysteamine to the gel-like formulation. The Ocular Surface Disease Index (OSDI) questionnaire[1] was applied before the switch and 3 months afterwards. The OSDI is a validated questionnaire, which is available for ocular surface disease (OSD), specifically for dry eye, and is designed to provide rapid assessment of the symptoms related to OSD and their impact on vision-related quality of life. Answers are graded from 0 (none of the time) to 4 (all of the time). Total score is calculated using the (sum of scores for all questions answered) x 100 / (total number of questions answered) x 4. This questionnaire addresses items over the past week including: sensitivity to light; gritty, painful eyes; blurred vision and limitations in reading; driving at night, working on a computer; being uncomfortable in windy conditions and areas that are air conditioned etc.

While two patients had an improvement in OSDI score, OSDI deteriorated for one patient, although this patient was known to have poor adherence to therapy. Sensitivity to light, sore eyes, blurred vision, and poor vision were the most common complaints reported. All patients felt that the 1-week stability of this new formulation was more convenient and improved quality of life. One patient felt that the consistency of the gel formulation constrained the eye drop installation.



- Corneal crystal deposition before starting topical cysteamine
- Visible reduction of corneal crystal deposition 6 months after starting cysteamine

Figure 1: Reduction of corneal cystine deposition after 6 months of cysteamine treatment.